
New RFLPs at the DXS164 (pERT 87-8) locus in the Black population

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SOURCE/DESCRIPTION: Probe pERT 87-8 is a 1.3 kb XbaI genomic fragment cloned in pUC18 (1).

CHROMOSOMAL LOCALIZATION: tightly linked to the Duchenne muscular dystrophy locus at Xp21 (1,2).

POLYMORPHISMS: In the Black population, probe pERT 87-8 recognizes a third allele in addition to the two seen by Kunkel et al. (1) when human genomic DNA is digested with either TaqI or BstXI (3). The new TaqI allele is 1.9 kb in length and the BstXI allele is 2.1 kb.

FREQUENCY: The frequencies of these alleles among 24 unrelated Blacks (15 females and 9 males) were:

| Allele | BstXI | | TaqI | |
|--------|----------|-----------|----------|-----------|
| | Size(kb) | Frequency | Size(kb) | Frequency |
| A1 | 4.4 | 0.03 | 1.1,2.7 | 0.64 |
| A2 | 2.2 | 0.73 | 3.8 | 0.08 |
| A3 | 2.1 | 0.24 | 1.9 | 0.28 |

NOT POLYMORPHIC FOR: No new RFLPs were detected with BamHI, PvuII, EcoRV, MspI, PstI, XmnI.

MENDELIAN INHERITANCE: Exhibited X-linked inheritance in 1 dystrophy family and 2 fragile X families.

PROBE AVAILABILITY: Contact Dr. L Kunkel.

OTHER COMMENTS: These alleles have recently been seen by another group (personal communication, J.F. Hejtmancik, Institute of Molecular Genetics, Baylor College of Medicine, Houston, TX) and by our group in samples supplied by K. Fischbeck (Dept. of Neurology, Univ. of Pennsylvania, Phila. PA).

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REFERENCES: 1. Monaco, AP et al. Nature 316:842-845, 1985; 2. Kunkel, LM et al. Nature 322:73-78, 1986; 3. Schwartz, C. et al., manuscript in preparation.